



isodicentric chromosome 15 syndrome

Isodicentric chromosome 15 syndrome is a developmental disorder with a broad spectrum of features. The signs and symptoms vary among affected individuals.

Poor muscle tone is commonly seen in individuals with isodicentric chromosome 15 syndrome and contributes to delayed development and impairment of motor skills, including sitting and walking.

Babies with isodicentric chromosome 15 syndrome often have trouble feeding due to weak facial muscles that impair sucking and swallowing; many also have backflow of acidic stomach contents into the esophagus (gastroesophageal reflux). These feeding problems may make it difficult for them to gain weight.

Intellectual disability in isodicentric chromosome 15 syndrome can range from mild to profound. Speech is usually delayed and often remains absent or impaired. Behavioral difficulties often associated with isodicentric chromosome 15 syndrome include hyperactivity, anxiety, and frustration leading to tantrums. Other behaviors resemble features of autistic spectrum disorders, such as repeating the words of others (echolalia), difficulty with changes in routine, and problems with social interaction.

About two-thirds of people with isodicentric chromosome 15 syndrome have seizures. In more than half of affected individuals, the seizures begin in the first year of life.

About 40 percent of individuals with isodicentric chromosome 15 syndrome are born with eyes that do not look in the same direction (strabismus). Hearing loss in childhood is common and is usually caused by fluid buildup in the middle ear. This hearing loss is often temporary. However, if left untreated during early childhood, the hearing loss can interfere with language development and worsen the speech problems associated with this disorder.

Other problems associated with isodicentric chromosome 15 syndrome in some affected individuals include minor genital abnormalities in males such as undescended testes (cryptorchidism) and a spine that curves to the side (scoliosis).

Frequency

Isodicentric chromosome 15 syndrome occurs in about 1 in 30,000 newborns.

Genetic Changes

Isodicentric chromosome 15 syndrome results from the presence of an abnormal extra chromosome, called an isodicentric chromosome 15, in each cell. An isodicentric chromosome contains mirror-image segments of genetic material and

has two constriction points (centromeres), rather than one centromere as in normal chromosomes. In isodicentric chromosome 15 syndrome, the isodicentric chromosome is made up of two extra copies of a segment of genetic material from chromosome 15, attached end-to-end. Typically this copied genetic material includes a region of the chromosome called 15q11-q13.

Cells normally have two copies of each chromosome, one inherited from each parent. In people with isodicentric chromosome 15 syndrome, cells have the usual two copies of chromosome 15 plus the two extra copies of the segment of genetic material in the isodicentric chromosome. The extra genetic material disrupts the normal course of development, causing the characteristic features of this disorder. Some individuals with isodicentric chromosome 15 whose copied genetic material does not include the 15q11-q13 region do not show signs or symptoms of the condition.

Inheritance Pattern

Isodicentric chromosome 15 syndrome is usually not inherited. The chromosomal change that causes the disorder typically occurs as a random event during the formation of reproductive cells (eggs or sperm) in a parent of the affected individual. Most affected individuals have no history of the disorder in their family.

Other Names for This Condition

- duplication/inversion 15q11
- idic(15)
- inv dup(15)
- inverted duplication 15
- isodicentric chromosome 15
- non-distal tetrasomy 15q

Diagnosis & Management

Other Diagnosis and Management Resources

- Autism Speaks: How is Autism Treated?
<https://www.autismspeaks.org/what-autism/treatment>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>

- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Chromosome
<https://medlineplus.gov/ency/article/002327.htm>
- Health Topic: Autism Spectrum Disorder
<https://medlineplus.gov/autismspectrumdisorder.html>
- Health Topic: Developmental Disabilities
<https://medlineplus.gov/developmentaldisabilities.html>
- Health Topic: Epilepsy
<https://medlineplus.gov/epilepsy.html>

Genetic and Rare Diseases Information Center

- Isodicentric chromosome 15 syndrome
<https://rarediseases.info.nih.gov/diseases/5153/isodicentric-chromosome-15-syndrome>

Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities
<https://www.genome.gov/11508982/>

Educational Resources

- MalaCards: isodicentric chromosome 15 syndrome
http://www.malacards.org/card/isodicentric_chromosome_15_syndrome
- March of Dimes: Chromosomal Conditions
<http://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Orphanet: Duplication/inversion 15q11
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3306
- Unique Rare Chromosome Disorder Support Group: Idic(15)
<http://www.rarechromo.org/information/Chromosome%2015/Idic%2815%29%20FTNW.pdf>

Patient Support and Advocacy Resources

- Chromosome Disorder Outreach
<http://chromodisorder.org/>
- Dup15q Alliance
<http://www.dup15q.org/understanding-dup15q/genetics-of-15q-duplications/types-of-15q-duplications/isodicentric-chromosome-15/>
- Unique Rare Chromosome Disorder Support Group
<http://www.rarechromo.org/html/home.asp>
- University of Kansas Resource List: Chromosomal Conditions
<http://www.kumc.edu/gec/support/chromoso.html>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28isodicentric+chromosome+15%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

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